

SLC22A5 gene

solute carrier family 22 member 5

Normal Function

The *SLC22A5* gene provides instructions for making a protein called OCTN2 that is found in the heart, liver, muscles, kidneys, and other tissues. This protein is positioned within the cell membrane, where it transports a substance known as carnitine into the cell. Carnitine is mainly obtained from the diet and is needed to bring certain types of fats (fatty acids) into mitochondria, the energy-producing centers within cells. Fatty acids are a major source of energy for the heart and muscles. During periods without food (fasting), fatty acids are also an important energy source for the liver and other tissues.

Health Conditions Related to Genetic Changes

Primary carnitine deficiency

More than 60 mutations in the *SLC22A5* gene have been found to cause primary carnitine deficiency. Some of these mutations create a premature stop signal in the instructions for making the OCTN2 protein, resulting in an abnormally short, nonfunctional protein. Other mutations change single protein building blocks (amino acids) in the OCTN2 protein.

Mutations in the *SLC22A5* gene result in an absent or dysfunctional OCTN2 protein. As a result, there is a shortage (deficiency) of carnitine within cells. Without carnitine, fatty acids cannot enter mitochondria and be used to make energy. Reduced energy production can lead to some features of primary carnitine deficiency, such as muscle weakness and hypoglycemia. Fatty acids may also build up in cells and damage the heart, liver, and muscles. This abnormal buildup causes the other signs and symptoms of the disorder.

Crohn disease

MedlinePlus Genetics provides information about Crohn disease

Other Names for This Gene

- CDSP

- high-affinity sodium dependent carnitine cotransporter
- novel organic cation transporter 2
- OCTN2
- organic cation transporter 5
- organic cation/carnitine transporter 2
- S22A5_HUMAN
- SCD
- solute carrier family 22 (organic cation transporter), member 5
- solute carrier family 22 (organic cation/carnitine transporter), member 5

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of SLC22A5 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6584\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6584[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28SLC22A5%5BTIAB%5D%29+OR+%28solute+carrier+family+22+member+5%29+OR+%28OCTN2%5BTIAB%5D%29+AND+%28carnitine+deficiency%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>)

Catalog of Genes and Diseases from OMIM

- SOLUTE CARRIER FAMILY 22 (ORGANIC CATION TRANSPORTER), MEMBER 5 (<https://omim.org/entry/603377>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6584>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SLC22A5\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SLC22A5[gene]))

References

- Amat di San Filippo C, Longo N. Tyrosine residues affecting sodium stimulation of carnitine transport in the OCTN2 carnitine/organic cation transporter. *J Biol Chem.* 2004 Feb 20;279(8):7247-53. doi: 10.1074/jbc.M309171200. Epub 2003 Dec 9. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14665638>)
- Amat di San Filippo C, Wang Y, Longo N. Functional domains in the carnitinetransporter OCTN2, defective in primary carnitine deficiency. *J Biol Chem.*

2003 Nov 28;278(48):47776-84. doi: 10.1074/jbc.M307911200. Epub 2003 Sep 23. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/14506273>)

- Inano A, Sai Y, Kato Y, Tamai I, Ishiguro M, Tsuji A. Functional regions of organic cation/carnitine transporter OCTN2 (SLC22A5): roles in carnitine recognition. Drug Metab Pharmacokinet. 2004 Jun;19(3):180-9. doi:10.2133/dmpk.19.180. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/15499185>)
- Koepsell H, Endou H. The SLC22 drug transporter family. Pflugers Arch. 2004 Feb; 447(5):666-76. doi: 10.1007/s00424-003-1089-9. Epub 2003 Jul 19. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12883891>)
- Lahjouji K, Mitchell GA, Qureshi IA. Carnitine transport by organic cationtransporters and systemic carnitine deficiency. Mol Genet Metab. 2001 Aug;73(4):287-97. doi: 10.1006/mgme.2001.3207. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/11509010>)
- Li FY, El-Hattab AW, Bawle EV, Boles RG, Schmitt ES, Scaglia F, Wong LJ. Molecular spectrum of SLC22A5 (OCTN2) gene mutations detected in 143 subjects evaluated for systemic carnitine deficiency. Hum Mutat. 2010 Aug;31(8): E1632-51.doi: 10.1002/humu.21311. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20574985>)
- Nezu J, Tamai I, Oku A, Ohashi R, Yabuuchi H, Hashimoto N, Nikaido H, Sai Y, Koizumi A, Shoji Y, Takada G, Matsuishi T, Yoshino M, Kato H, Ohura T, Tsujimoto G, Hayakawa J, Shimane M, Tsuji A. Primary systemic carnitine deficiency is caused by mutations in a gene encoding sodium ion-dependent carnitinetransporter. Nat Genet. 1999 Jan;21(1):91-4. doi: 10.1038/5030. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/9916797>)
- Tein I. Carnitine transport: pathophysiology and metabolism of known molecular defects. J Inherit Metab Dis. 2003;26(2-3):147-69. doi: 10.1023/a:1024481016187. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/12889657>)

Genomic Location

The *SLC22A5* gene is found on chromosome 5 (<https://medlineplus.gov/genetics/chromosome/5/>).

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